

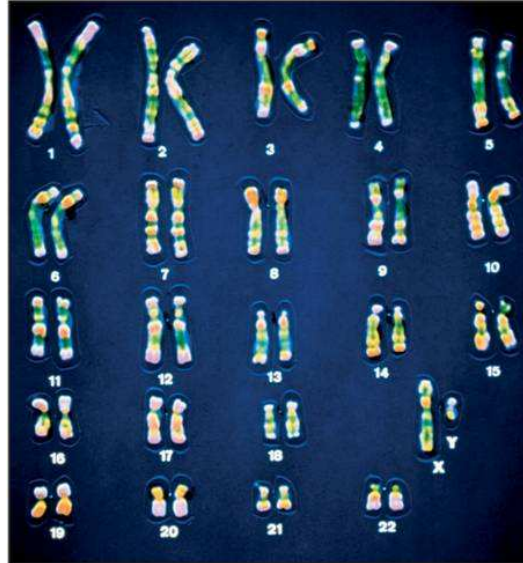
# Karyotypes and Nondisjunction

A **karyotype** is a picture of human chromosomes arranged by homologous pairs.

Cells are photographed during metaphase of mitosis. Scientists then cut out the chromosomes from the photographs and group them together in pairs.

Cell biologists use karyotypes to identify chromosomal abnormalities.

Normal number is 46. Abnormal is 45, 47, 48 or more.



Human Karyotype – chromosomes come from cell's in the amniotic fluid which surrounds the baby. Removed through *amniocentesis*.

Two of the 46 human chromosomes are known as **sex chromosomes**, because they determine an individual's sex.

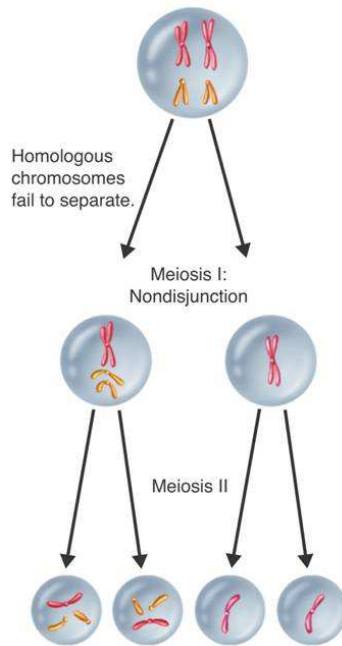
- Females have two copies of an X chromosome.
- Males have one X chromosome and one Y chromosome.

The remaining 44 chromosomes are known as autosomal chromosomes, or **autosomes**.

## Chromosomal Disorders

- The most common error in meiosis occurs when homologous chromosomes fail to separate.
- This is known as **nondisjunction**, which means, “not coming apart.”

If nondisjunction occurs, abnormal numbers of chromosomes may find their way into gametes, and a disorder of chromosome numbers may result.



If two copies of an autosomal chromosome fail to separate during meiosis, an individual may be born with three copies of a chromosome.

**Down Syndrome** involves three copies of chromosome 21. “**Trisomy 21**”

Down Syndrome produces mild to severe mental retardation.

It is characterized by:

- increased susceptibility to many diseases
- higher frequency of some birth defects
- Recognizable facial features

### Sex Chromosome Disorders

In females, nondisjunction can lead a female which usually inherits only one X chromosome (karyotype = 45, XO “**monosomy**”).

**Turner’s Syndrome** these women are sterile, do not develop normally, short thick webbed neck, some mild retardation.

Can also have 3 XXX at pair #23. these are females, some develop normally, some are sterile.

In males, nondisjunction causes an extra X chromosome (karyotype 47,XXY)

**Klinefelter’s Syndrome** males, sterile, some degree of mental retardation.

There can also be extra Y chromosomes

at pair # 23 XYY or XYYY.

**Extra chromosomes can end up at different autosomal positions also.**